HSD3B7 gene

hydroxy-delta-5-steroid dehydrogenase, 3 beta- and steroid delta-isomerase 7

Normal Function

The *HSD3B7* gene provides instructions for making an enzyme called 3 beta-hydroxysteroid dehydrogenase type 7 (3β-HSD7). This enzyme is found in liver cells. It is embedded in the membrane of a cell structure called the endoplasmic reticulum, which is involved in protein processing and distribution. The 3β-HSD7 enzyme participates in the production of bile acids, which are a component of a digestive fluid called bile. Bile acids stimulate bile flow and helps absorb fats and fat-soluble vitamins. Bile acids are produced from cholesterol in a multi-step process. The 3β-HSD7 enzyme is responsible for the second step in that process, which converts 7alpha(α)-hydroxycholesterol to 7α-hydroxy-4-cholesten-3-one.

Health Conditions Related to Genetic Changes

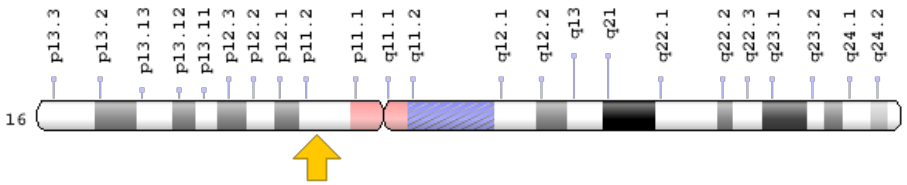
congenital bile acid synthesis defect type 1

At least 17 mutations in the *HSD3B7* gene have been found to cause congenital bile acid synthesis defect type 1. This condition is characterized by cholestasis, a condition that impairs the production and release of a digestive fluid called bile from liver cells. Most of the *HSD3B7* gene mutations delete one or two DNA building blocks (base pairs) from the gene or replace single protein building blocks (amino acids) in the enzyme. These mutations result in production of a 3β-HSD7 enzyme with little or no function. Without enough functional 3β-HSD7 enzyme, the conversion of 7α-hydroxycholesterol to 7α-hydroxy-4-cholesten-3-one is impaired. The 7α-hydroxycholesterol instead gets converted into abnormal bile acid compounds that cannot be transported out of the liver into the intestine, where the bile acids are needed to absorb fats and fat-soluble vitamins. This impaired production and release of bile acids leads to cholestasis. As a result, cholesterol and abnormal bile acids build up in the liver and fat-soluble vitamins are not absorbed, leading to the signs and symptoms of congenital bile acid synthesis defect type 1.

Chromosomal Location

Cytogenetic Location: 16p11.2, which is the short (p) arm of chromosome 16 at position 11.2

Molecular Location: base pairs 30,985,189 to 30,989,152 on chromosome 16 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 3-beta-HSD VII
- 3 beta-hydroxy-delta 5-C27-steroid oxidoreductase
- 3-beta-hydroxy-Delta(5)-C27 steroid oxidoreductase
- 3 beta-hydroxysteroid dehydrogenase type 7
- 3 beta-hydroxysteroid dehydrogenase type VII
- 3BHS7_HUMAN
- c(27) 3-beta-HSD
- C(27)-3BETA-HSD
- cholest-5-ene-3-beta,7-alpha-diol 3-beta-dehydrogenase
- SDR11E3
- short chain dehydrogenase/reductase family 11E, member 3

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: Fat Absorption and Lipid Metabolism in Cholestasis
  https://www.ncbi.nlm.nih.gov/books/NBK6420/
- Madame Curie Bioscience Database: Overview of Biliary Anatomy and Morphology
  https://www.ncbi.nlm.nih.gov/books/NBK6407/#A27298

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28HSD3B7%5BTIAB%5D%29+OR+%283+beta-hydroxy-delta+5-C27-steroid+oxidoreductase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

OMIM

- 3-BETA-HYDROXY-DELTA-5-C27-STEROID OXIDOREDUCTASE
  http://omim.org/entry/607764

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=HSD3B7%5Bgene%5D
- HGNC Gene Family: Short chain dehydrogenase/reductase superfamily
  http://www.genenames.org/cgi-bin/genefamilies/set/743
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=18324
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/80270
- UniProt
  http://www.uniprot.org/uniprot/Q9H2F3

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